MATN3 gene

matrilin 3

Normal Function

The *MATN3* gene provides the instructions for making a protein called matrilin-3. This protein is found in the extracellular matrix, which is an intricate lattice of proteins and other molecules that forms in the spaces between cells. Specifically, matrilin-3 is located in the extracellular matrix surrounding the cells that make up ligaments and tendons, and near cartilage-forming cells (chondrocytes). Chondrocytes play an important role in bone formation (osteogenesis). In the bones of the spine, hips, and limbs, the process of osteogenesis starts with the formation of cartilage, which is then converted into bone.

The normal function of the *MATN3* gene is not fully understood; however, research suggests that matrilin-3 may play a role in the organization of collagen and other cartilage proteins. Collagens are proteins that provide strength and support to many body tissues, including cartilage. Matrilin-3 has been shown to interact with the COMP protein, type II collagen, and type IX collagen, which are all important in cartilage and bone formation.

Health Conditions Related to Genetic Changes

multiple epiphyseal dysplasia

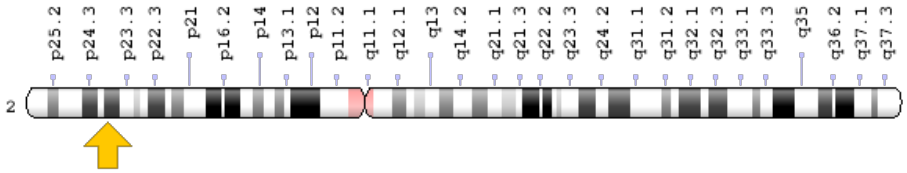
At least 14 different mutations in the *MATN3* gene have been shown to cause a mild form of multiple epiphyseal dysplasia. All of the mutations change one protein building block (amino acid) within or close to a region of matrilin-3 called the A-domain. One genetic change accounts for approximately 40 percent of all *MATN3* mutations. This mutation replaces the amino acid arginine with the amino acid tryptophan at position 121 (written as Arg121Trp or R121W).

Researchers believe that mutations in the *MATN3* gene prevent matrilin-3 from folding properly. Instead of being transported to the extracellular matrix of the chondrocytes, matrilin-3 remains in the endoplasmic reticulum. The endoplasmic reticulum is a structure inside the cell that is involved in protein processing and transport. This cell structure eventually becomes so large that it is no longer able to function normally, and the chondrocyte dies. The premature death of chondrocytes results in diminished growth of the long bones and short stature.

Chromosomal Location

Cytogenetic Location: 2p24.1, which is the short (p) arm of chromosome 2 at position 24.1

Molecular Location: base pairs 19,992,052 to 20,012,694 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- EDM5
- MATN3_HUMAN

Additional Information & Resources

GeneReviews

- Multiple Epiphyseal Dysplasia, Dominant
  https://www.ncbi.nlm.nih.gov/books/NBK1123

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MATN3%5BTIAB%5D%29+OR+%28matrilin+3%5BTIAB%5D%29%29+OR+%28EDM5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- MATRILIN 3
  http://omim.org/entry/602109

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MATN3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MATN3%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6909
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4148
- UniProt
  http://www.uniprot.org/uniprot/O15232

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